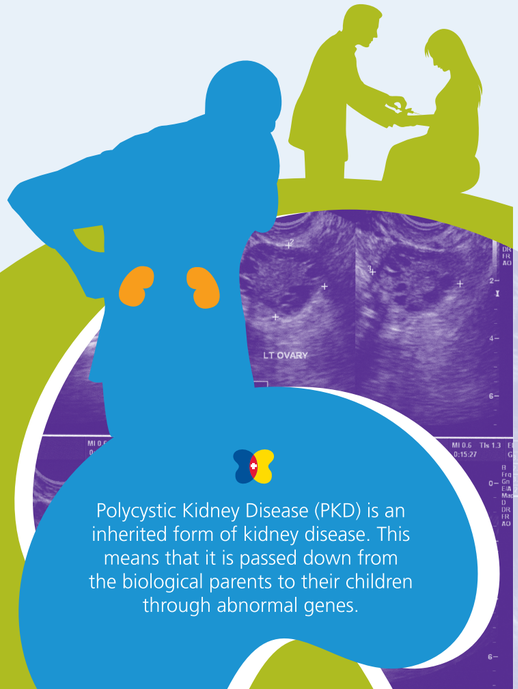
KDF
KIDNEY DIALYSIS FOUNDATION

POLYCYSTIC KIDNEY DISEASE



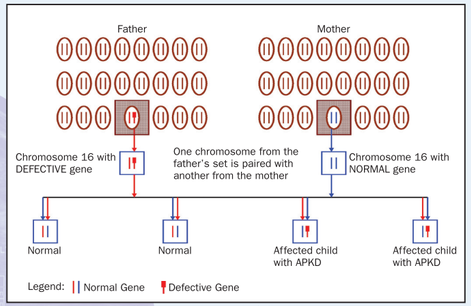
Polycystic Kidney Disease (PKD) is an inherited form of kidney disease. This means that it is passed down from the biological parents to their children through abnormal genes.

Polycystic Kidney Disease

What is Polycystic Kidney Disease?

Polycystic Kidney Disease (PKD) is an inherited form of kidney disease. This means that it is passed down from the biological parents to their children through abnormal genes. It was first described in the 19th century and many polycystic kidney disease families have been identified. There are 2 forms of this disease: the adult form which we will call Adult Polycystic Kidney Disease (APKD) and the rarer Infantile Polycystic Kidney Disease (IPKD).

APKD and IPKD are inherited differently – the APKD form is inherited in a **dominant** fashion i.e. if **one** parent has the defective gene in one of the two sets of 23 chromosomes and the gene is passed to his/her child the disease will manifest itself. Therefore, there is a 50% chance of the defective gene being passed on. This is entirely random and a parent can have 4 children and lucky enough to pass on only the set which contains the normal gene, so none of his children are affected. The disease may appear to skip a generation if the affected parent died young without showing some of the signs and symptoms.



Note: - 50% chance that the defective gene will be passed onto the child.

In the early stage of the disease, many people do not know they are affected if they have no complaints and did not have an ultrasound scan performed. We now know that there are 3 types of genes involved in APKD (PKD1 on chromosome 16, PKD2 on chromosome 4 and PKD3 the location of which is unclear as yet).

On the other hand, in the Infantile form (IPKD), the inheritance is **recessive**. It requires **both** parents to pass the defective gene to their child before the disease manifests itself.

Renal Failure and APKD

Adult Polycystic Kidney Disease (APKD), is the more common variety. Its incidence has been quoted to be about 1 in 1000 persons.

APKD affects the nephron. The nephron is the filtering unit of the kidneys and we have about 1 million of them in each kidney. The nephron comprises the filter (or glomerulus) and the tubule which drains the fluid that has been filtered into the ureter. The drained fluid is collected in the bladder to be passed out as urine.

In APKD, cysts form along the tubule. In time to come, compression of normal tissue and a scarring reaction occurs. The scarring reaction results from certain substances being secreted by the injured kidney. This process also involves a reduction of the blood supply in the very small vessels of the kidney further damaging the kidney.

The rate at which this occurs is different for every affected individual. Most of the affected people do not develop renal failure until they are in their 40's. Some may even be affected only in their 60's or 70's.

What are the symptoms and signs of the disease?

- **Cysts in the kidneys**
  In the early stage there are no symptoms and the disease is usually picked up when ultrasound scanning is done for other reasons and cysts are noted in the kidneys. Health screening may pick up previously undetected high blood pressure or kidney failure.

- **Enlarged Abdomen**
  As the cysts in the kidneys enlarge, patients may notice their abdomen getting bigger. The enlarging kidneys may cause some vague discomfort. There may be more severe pain if there is an infection involving the cysts or bleeding into the cysts. Infections of polycystic kidneys require more specific antibiotics than infections in normal kidneys because of the presence of cysts. The duration of treatment is also longer and usually last for several weeks.

- **Blood in Urine**
  Bleeding from cyst ruptures may also cause episodes of passing urine with blood. This may be aggravated by trauma (physical injury) so patients should not engage in body-contact sports. Bleeding is usually self limiting. Only rarely does the kidney needs to be removed because of persistent bleeding and severe blood loss.

- **Hypertension**
  Many affected individuals develop hypertension. Uncontrolled hypertension can hasten the progression of renal failure. It also has effects on the heart and is a risk factor in the development of heart failure, heart attack and stroke.

Are other organs affected in this disease?

Cysts can also develop in the liver as well as the pancreas. These do not usually cause problems.

Patients with APKD may have a higher incidence of :

- Aneurysms (outpouching of the blood vessels) in the brain which may rupture
- Prolapse mitral valve (abnormality of one of the heart valves)
- Colonic diverticulosis (outpouchings of the large intestine)
- Kidney stones

Can kidney failure be prevented?

- **Blood pressure control**
  To delay the onset of kidney failure, very good blood pressure control is necessary. There are many blood pressure medications and your doctor will help you find a suitable one.

- **Restriction of protein and salt intake**
  A restricted protein diet also helps to retard the progression of kidney failure. However, too strict a diet may cause malnutrition and this should be supervised by a dietician. Remember that restriction of salt is important in helping to maintain good blood pressure control.

What happens when the kidneys have totally failed?

As mentioned earlier, it may take many years before the kidneys totally fail. Dialysis will be needed to sustain life. If the patient is suitable for transplantation, renal transplantation may be a better option. Before this happens, you should try to learn more about dialysis and transplantation from your doctor and other medical sources. A significant amount of preparation is required for a smooth transition to dialysis without the need for long periods of hospitalization. This is also important for people who are working so that there is minimal interruption to work.

Who should be tested for APKD?

All the direct blood relatives of an affected individual are at risk of having APKD and should be given advice regarding this illness. Because of the possibility of renal failure and the effects of high blood pressure, they should be encouraged to go for screening. The most convenient and harmless method of screening for APKD is by ultrasound scanning. Most cysts will be evident by the time an individual is 30 years old.

Can women with APKD have children?

Patients with APKD can undergo normal pregnancies. There is increased risk if hypertension is already present or if renal function is already abnormal.

Patients who are affected with APKD (both men and women) should consider the fact that each of their children have a 50% chance of the abnormal gene being passed to them. However, almost all children will live to adulthood.

If you have any further questions, you should approach your doctor who will be able to address your concerns.

You may visit these websites for more information:

http://www.kdf.org.sg/health.aspx
http://www.davita.com
http://www.uptodate.com/patients/index.html

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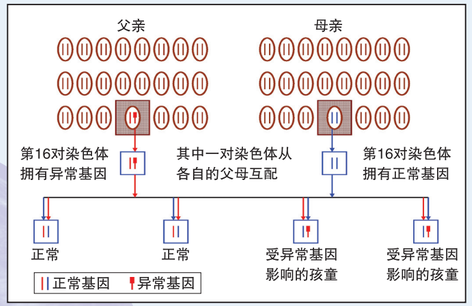
多囊肾病

早在19世纪，多囊肾病已被证实是由父亲或母亲的异常因子遗传给后代而形成的一种遗传性肾疾病。

什么是多囊肾病？

早在19世纪，多囊肾病已被证实是由父亲或母亲的异常因子遗传给后代而形成的一种遗传性肾疾病。它一般可分为两大类：在成年后发病的，称为成人多囊肾病（APKD），另一类则是较少见的幼儿多囊肾病（IPKD），两者的遗传机理是不同的。

成人多囊肾病的遗传型式是显性的，父母各自因子中的23对染色体，只要有一对是异常的，即有可能导致子女发病。疾病的遗传机率是50%，且没有固定性的发病期。例如一位父/母有四名子女，他/她有可能很幸运地只把正常的因子遗传给后代，结果没有一名子女受到异常因子的影响。再者，这遗传的延续也会因为患者的早逝，未被诊断出来，而出现间断的现象。



注意：异常因子被遗传下一代有50%的机率。

初期的成人多囊肾病是不容易被察觉的，患者一般也与常人无异，须要通过超音波扫描，才能验证。目前，医学上发现会影响成人多囊肾病的染色体定位有：PKD1 – 第16对染色体异常；PKD2 – 第4对染色体异常；PKD3 – 尚无染色体定位。幼儿多囊肾病的遗传则是隐性的，只有在父母两人都带有异常因子并同时遗传给子女，才有患上这疾病的可能性。

我们将着重详述成人多囊肾病，因为这是较常见的病种，其发生率是每一千人当中就有一名患者。

多囊肾病如何导致肾衰竭？

成人多囊肾病会损害肾单位的结构。肾单位是肾脏的基本功能单位，每颗肾脏有超过100万个肾单位，每个肾单位是由肾小球（过滤器）和肾小管所组成的。它的功能是帮助过滤和排除水分，通过输尿管收集到膀胱，然后排出体外。

成人多囊肾病首先会在肾小管上形成囊泡，经过一定的时间后，肾小球的正常组织就会因受到压迫而出现疤痕，导致某些物质从受损的肾脏排出，同时也降低肾脏微血管的血液供应，加重肾脏的损伤。

肾脏损伤的进度是因人而异的，大多数患者的肾功能在40岁以后才开始衰竭，有些则能延缓到60岁或甚至70岁以上。

成人多囊肾病有什么症状？

- **肾囊泡**
  初期没有明显的症状，通常是患者在为其他疾病进行超音波扫描时才被发现。从全身的健康检验中也可察觉以往所未曾发现的高血压或肾衰竭。

- **腹部肿胀**
  当肾囊泡逐渐肿大，患者会因腹部的肿胀而感到轻微不适。如果肾囊泡受到感染或出血，患者就会有剧烈的疼痛。此外，患者必须服用分量比普通感染病更重的特定消炎药，而且也需要更长的治疗时间，一般需要几个星期。
- **血尿**
  肾囊泡破裂可能会造成血尿，外伤的程度也可能会加剧，因此患者不宜参与撞击性的运动（如：足球、跆拳道等）。然而，出血的现象一般都会自行停止，因出血不止或失血过多而必须切除肾脏的事例是罕有的。

- **高血压**
  多囊肾病患者通常会患有高血压，如果没有良好控制，将会加速肾衰竭的进度。高血压也会影响心脏，引发心脏病、中风或心功能衰竭。

多囊肾病是否会影响其他器官？

囊泡也会在肝脏和胰脏内滋长，但这些囊泡一般不会造成问题。

患成人多囊肾病的人士容易患上以下疾病：

- 脑血管瘤 (可能会破裂)
- 二尖瓣脱垂（异常的心脏瓣膜）
- 结肠憩室炎
- 肾结石

肾衰竭可否预防？

- **控制血压**
  良好的血压控制有助于延缓肾衰竭，医生会协助患者找出适合的高血压抑制药物。

- **限制蛋白质和盐分的摄取**
  限制蛋白质的摄取是延缓肾衰竭的方法之一，但过度的控制也会造成营养不良，因此营养师会监督患者的饮食习惯。限制盐分的摄取则有助于血压的控制，日常的饮食中一般都隐藏着很多的盐分，所以必须小心控制。

肾衰竭到达末期后，需要什么治疗？

肾功能的衰竭可能在数年后才会到达末期，到时患者必须依靠洗肾治疗来维持生命。对于适合接受肾脏移植治疗的患者，移植治疗可能是更好的方法。

在肾衰竭还未到达末期前，患者应该向医生和有关的医药资讯机构了解洗肾与肾脏移植治疗的程序。有了充分的准备，患者就可以很顺利地接受洗肾治疗，不必长期住院，大大减少对工作的影响，这对有工作的肾病患者是非常重要的。

那些人应该接受成人多囊肾病的检验？

所有多囊肾病患者的直系家属都有患上这疾病的可能性，都应该接受检验。此外，他们患上肾衰竭和高血压的机率也很高，所以都应该接受例常检查。通常囊泡会在患者约30岁时出现，最普遍而又无副作用的检查方法是超音波扫描。

患有成人多囊肾病的妇女可否生育？

患有这疾病的妇女是可以怀孕的，但如果患者的肾功能已出现异常现象及患上高血压，这将增加生育的危险性。

患有成人多囊肾病的父/母，都应该意识到他/她的子女都有50%的异常因子遗传机率，然而，多数患者的子女即使带有异常基因，也能长大成人。

若有任何疑问，应该向您的医生询问，他会更了解您的需要。

欲知详情，可参阅以下的网址：

http://www.kdf.org.sg/health.aspx
http://www.davita.com
http://www.uptodate.com/patients/index.html

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